HIGHER ANEUPLOIDY RISK IN SPERM AND ABNORMAL MEIOTIC RECOMBINATION LEVELS IN NON-OBSTRUCTIVE AZOOSPERMIA PATIENTS

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Objective: The aim of this study was to asses the correlation between meiotic recombination and the incidence of chromosomal abnormalities in testicular spermatozoa from azoospermic infertile males.

Material and methods: From January 2008-December 2010, meiotic recombination in pachytene stage cells and sperm aneuploidy were evaluated in samples obtained from testicular biopsies of 11 non-obstructive azoospermic patients (NOA). The study group was compared with a control group formed of 10 post-vasectomy patients (OA). Meiotic recombination was assessed using immunocytogenetics with three primary monoclonal antibodies: anti-SCP3 directed to axial/lateral elements of the synaptonemal complex; CREST directed to the centromeres of chromosomes and a MutL homolog1 antibody (MLH1), which is a mismatch repair protein that co-localizes to sites of meiotic cross-overs. Aneuploidy rates on sperm for chromosomes 13, 18, 21, X, and Y were analyzed by fluorescence “in situ” hybridization (FISH).

Results: The overall mean number of MLH1 foci per cell showed a significant decrease in NOA compared to the control group of post-vasectomized patients (44.9±3.6 vs. 48.2 ± 2.1, $p<0.0001$). FISH analysis revealed a two-fold increase for sex chromosomes disomies in NOA patients (0.39 vs. 0.18; $p=0.0003$) and a three-fold increase for chromosome 13 (0.4 vs. 0.09; $p<0.0001$), chromosome 21 (0.3 vs. 0.09; $p=0.0025$) and diplody rates (0.13 vs. 0.05; $p= 0.0029$).

Conclusions: Our study shows a decrease in recombination levels and increased sperm aneuploidy rates in NOA patients. These findings would corroborate the correlation between both parameters and the higher aneuploidy risk for the offspring of NOA patients.